Office of Public Health Genomics
Evaluating Genomic Tests and Family History

Identifying Opportunities to Improve Health and Transform Healthcare

CDC's Office of Public Health Genomics (OPHG) is working to integrate advances in genomics effectively and responsibly into public health programs to improve population health.

A Tiered Approach to Integrating Genomic Tests and Family Healthy History into Practice

In 2012, OPHG developed a three-tiered framework for classifying genomic testing and family health history applications based on the availability of scientific evidence and evidence-based recommendations supporting their use.

Table. Evidence-based Classification of Genomic Tests and Family Health History

Tier	Definition	Example(s)
1	Implementation in practice supported by base of synthesized evidence.	*BRCA*-associated hereditary breast and ovarian cancer (US Preventive Services Task Force B recommendation); Lynch syndrome (EGAPP)
2	May provide information for informed decision making based on existing evidence; however, synthesized evidence is insufficient to support routine implementation in practice.	Family health history in primary care, with few exceptions
3	Not ready for routine implementation in practice based on synthesized evidence culminating in recommendations against use, OR no relevant synthesized evidence identified.	Direct-to-consumer personal genomic tests

The Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group

The EGAPP Working Group (EWG) is an independent, non-federal, multidisciplinary panel, supported by OPHG since 2005. The EWG has published systematic methods to assess the validity and utility of genomic testing and family health history applications; and nine recommendation statements on genomic applications for breast cancer, colorectal cancer, prostate cancer, heart disease, diabetes and depression.

EGAPP: Informing Policy at the National, State, and Institutional Level

- *Healthy People 2020*: The 2009 EGAPP recommendation on genetic testing for Lynch syndrome served as the foundation for a new developmental objective in the genomics topic area.
- *Payer coverage:* Major health plans, including Blue Cross Blue Shield licensees in several states, are basing policies on the 2009 EGAPP recommendation on genetic testing for Lynch syndrome.
- *Institutional screening protocols*: More than 90 institutions have used the 2009 EGAPP Lynch syndrome recommendation to justify Lynch syndrome screening protocols.

Next Steps

- For 2014, the EWG is preparing two new recommendations statements on genomic applications for prostate cancer risk assessment and antiplatelet therapy; as well as new methods for evidence synthesis and modeling, including stratified screening.
- Through ongoing horizon scanning, OPHG continues to build a database of genomic applications in transition from research to clinical and public health practice, numbering over 500 since 2009. (http://www.hugenavigator.net/GAPPKB/home.do)
- OPHG continues to conduct horizon scanning for the very few evidence-based reviews and recommendations on genomic tests and family health history from other sources, upon which to base public health programs and policy.
- OPHG is actively exploring sustainable and scalable models for supporting the EWG, or an EGAPP-like process.

More Information

Office of Public Health Genomics
Centers for Disease Control and Prevention

1600 Clifton Road NE
Mailstop E-61
Atlanta, GA 30333

800-CDC-INFO (800-232-4636)
TTY: (888) 232-6348

www.cdc.gov/genomics/
cdcinfo@cdc.gov

Center for Surveillance, Epidemiology and Laboratory Services
Office of Public Health Genomics

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